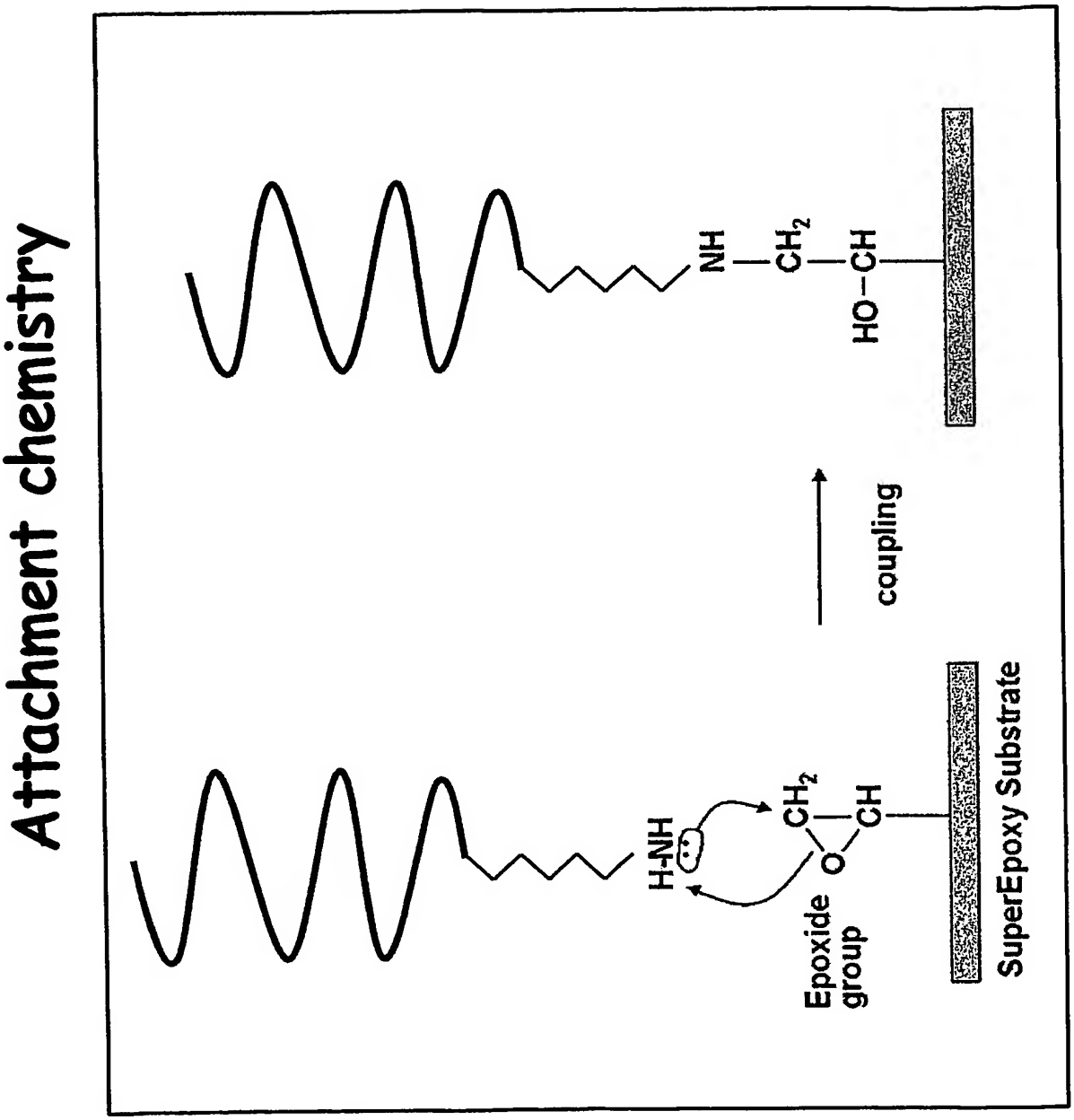

Attachment chemistry
H-NH
CH2
CH
O
Epoxide group
coupling
NH
CH2
CH
HO
SuperEpoxy Substrate

Figure 1

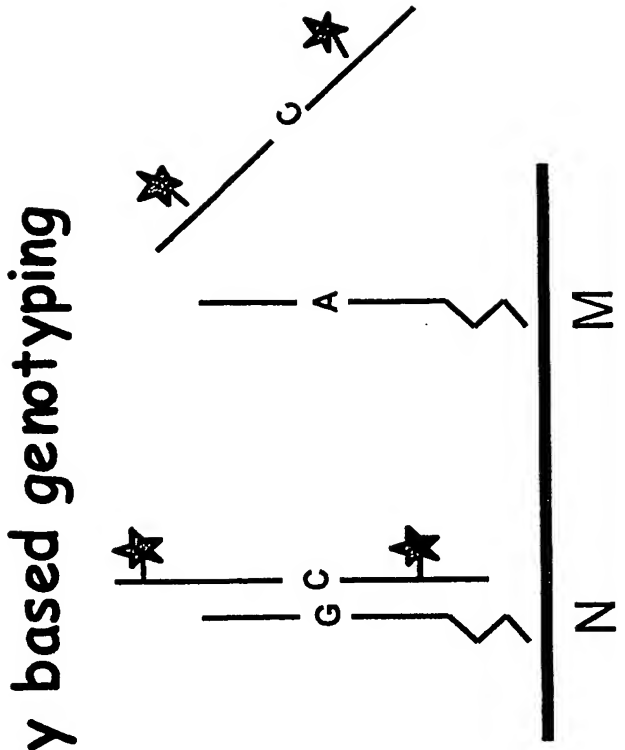

Microarray based genotyping
Allele specific
oligonucleotides (ASO)
G
C
A
C
N
M

Figure 2

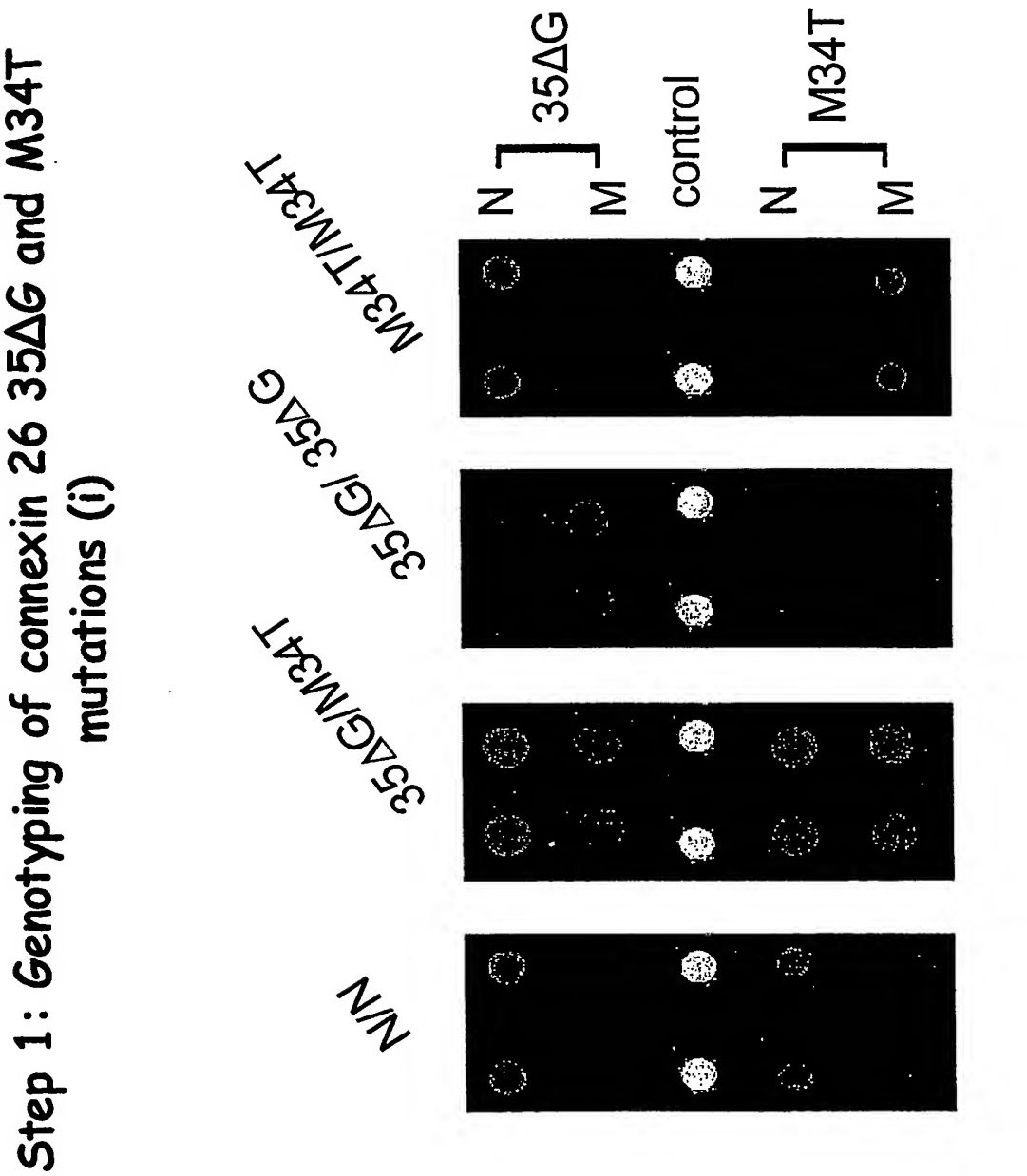

Step 1: Genotyping of connexin 26 35ΔG and M34T mutations (i)
N/N
35AG/M34T
35AG/ 35AG
M34T/M34T
N
M
35ΔG
control
N
M
M34T

Figure 3

Step 1: Genotyping of connexin 26 35ΔG and M34T mutations (ii)

$$\text{Genotype Index (GI)} = \frac{SV_N}{(SV_N + SV_M)}$$

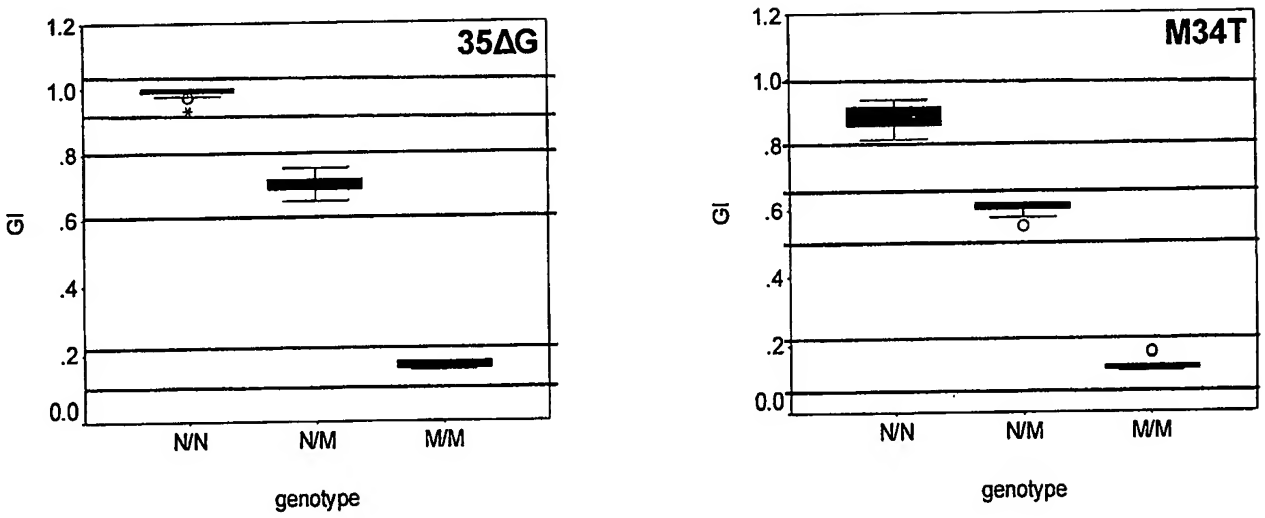


Figure 4

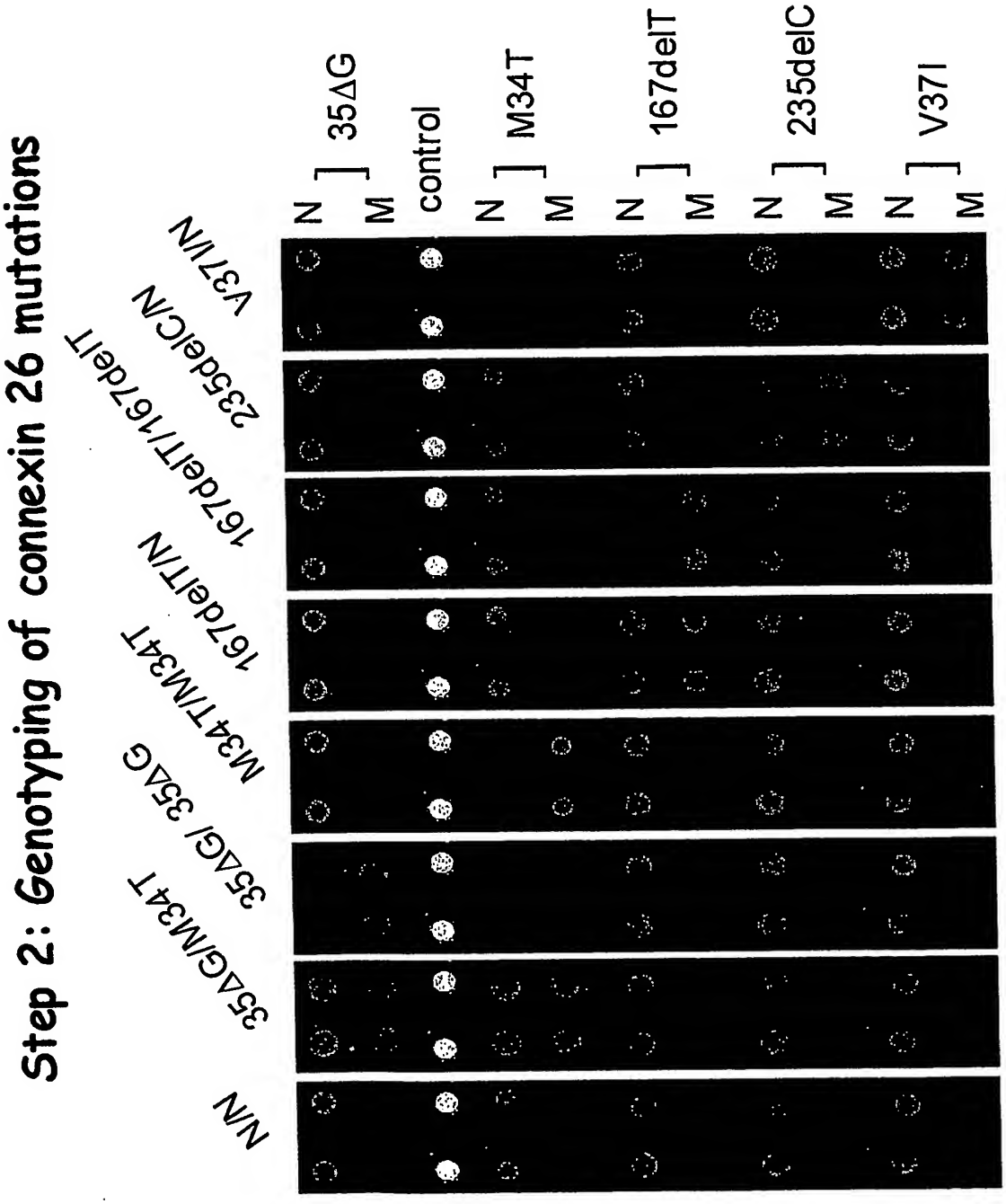

Step 2: Genotyping of connexin 26 mutations
N/N
35AG/M34T
35AG/ 35AG
M34T/M34T
167delTT/N
167delTT/167delTT
235delC/N
V37I/N
N
M
control
35ΔG
M34T
167delT
235delC
V37I

Figure 5

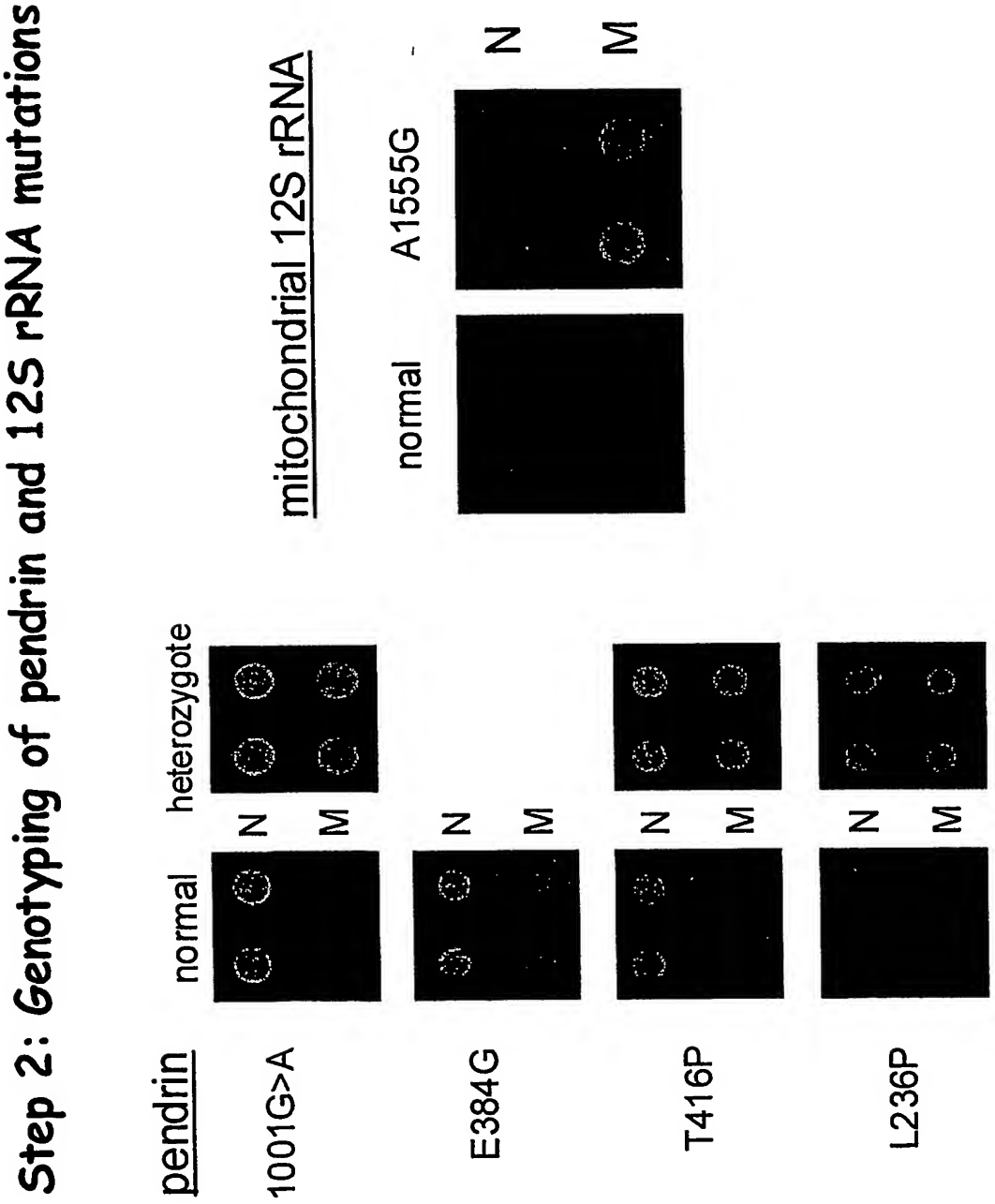

Step 2: Genotyping of pendrin and 12S rRNA mutations
pendrin
normal
heterozygote
N
M
1001G>A
E384G
T416P
L236P
mitochondrial 12S rRNA
normal
A1555G
N
M

Figure 6

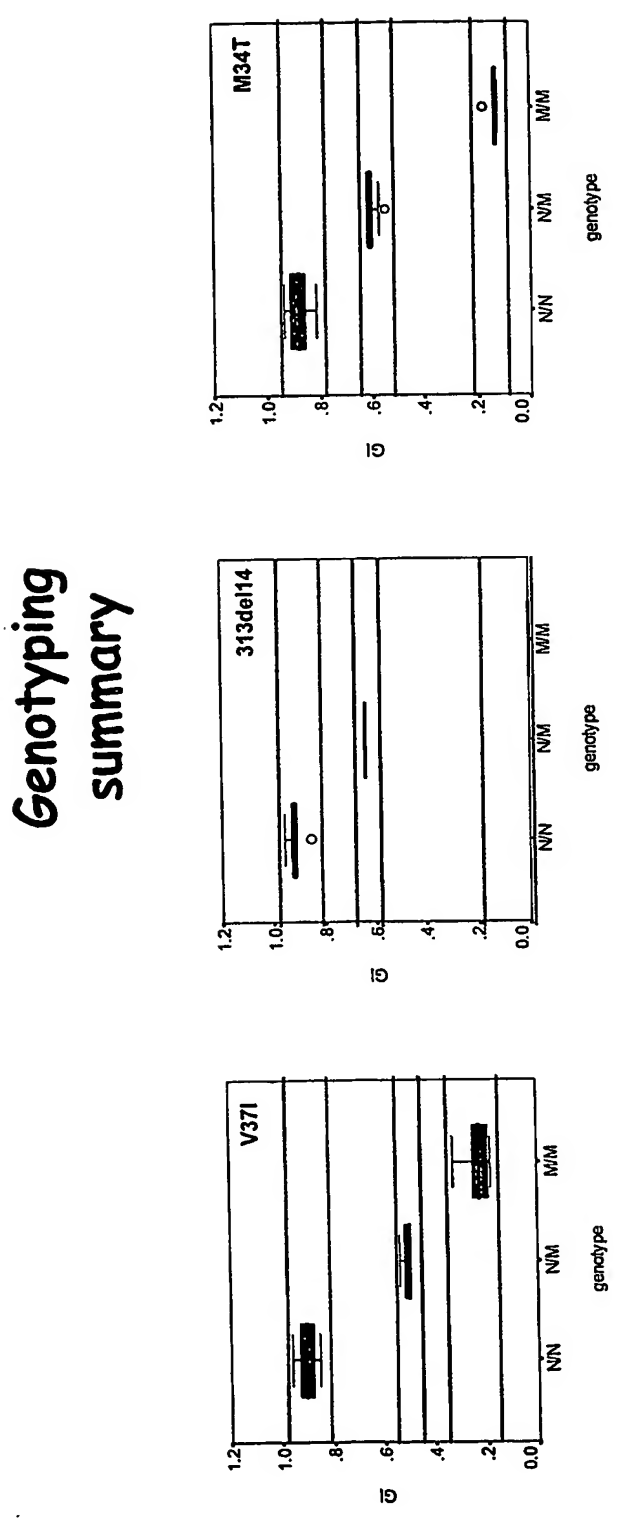


Figure 7A

Figure 7B

Figure 7C

Figure 7D

Figure 7E

Figure 7F

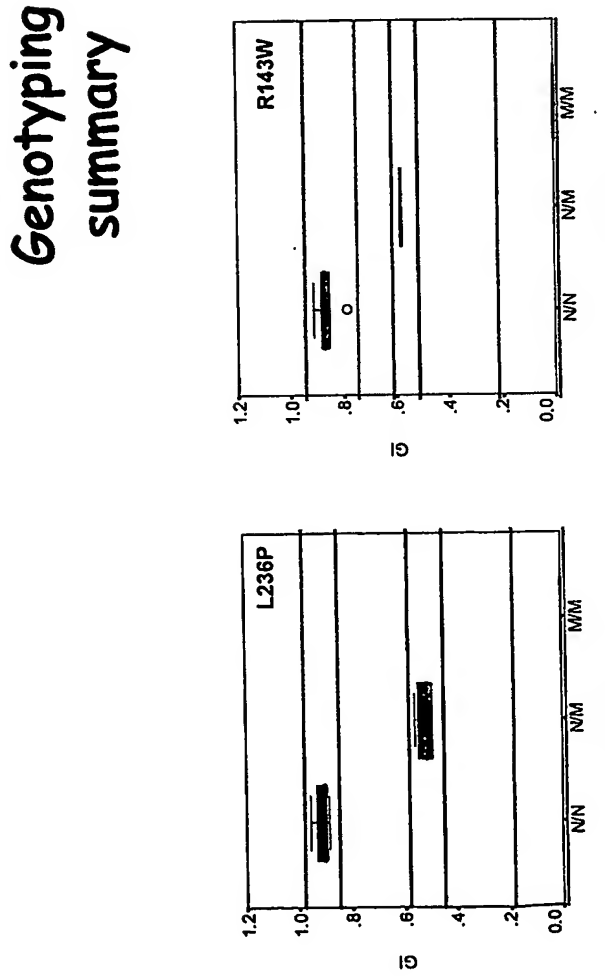


Figure 7G

Figure 7I

Figure 7J

Figure 7H

Figure 7K

Figure 7L

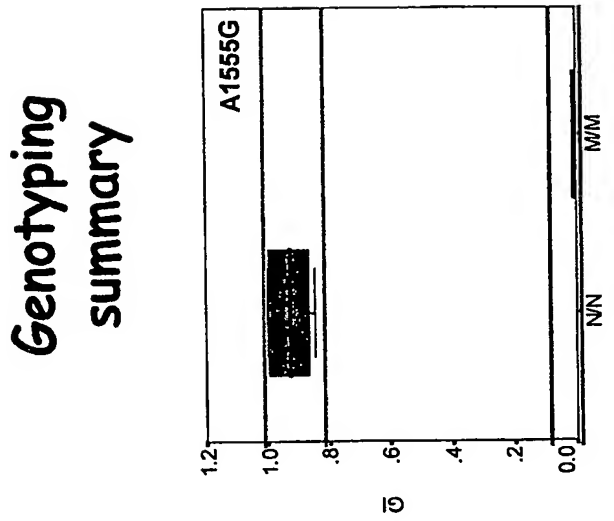


Figure 7M

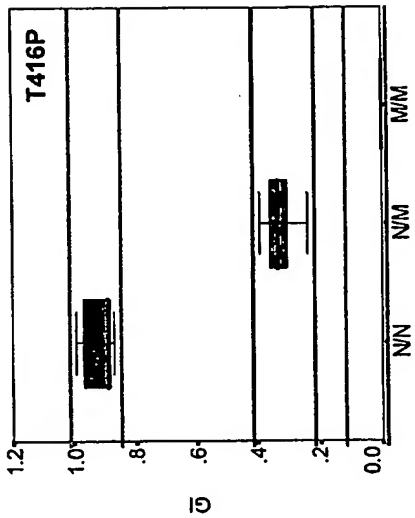
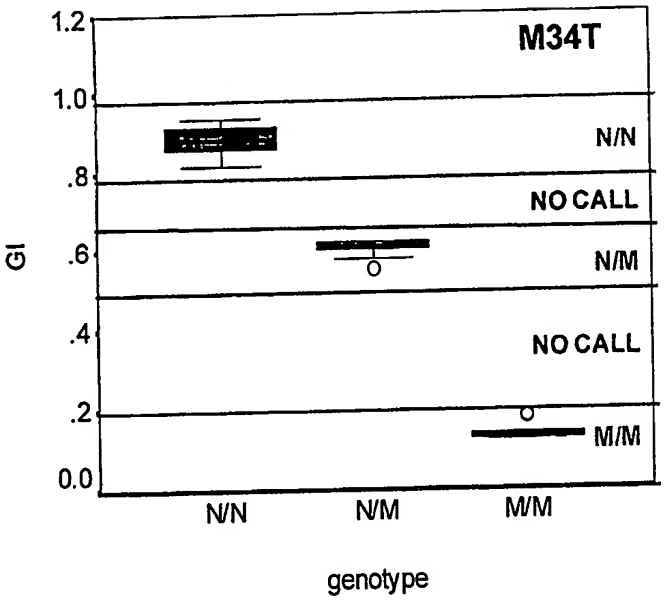


Figure 7N

Genotype calling algorithm



If 0.8<GI<1.0 then call = N/N

If 0.65<GI<0.8 then call = NO CALL

If 0.65<GI<0.5 then call = N/M

If 0.2<GI<0. 5 then call = NO CALL

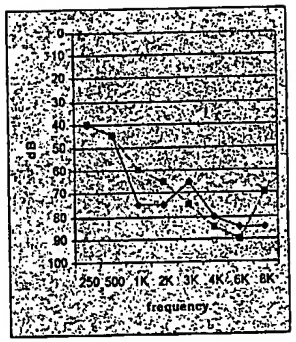
If 0.0<GI<0.2 then call = M/M

If $SI_N+SI_M<100$ then call = NO CALL

Figure 8

Interactions between deafness genes?

- severe hearing loss
- progressive
- age of onset 10 years



Mutation	GI	Call
connexin 26		
35delG	0.992806	N/N
W24X	0.93836	N/N
M34T	0.619185	**N/M**
V37I	0.902981	N/N
167delT	0.999151	N/N
235delC	0.997346	N/N
L90P	0.992122	N/N
R143W	0.862635	N/N
313del14	0.932165	N/N

Mutation	GI	Call
pendrin		
L236P	0.935414	N/N
G1001A	0.906897	N/N
E384G	0.941176	N/N
T416P	0.969543	N/N
12S rRNA		
A1555G	0.007764	**M/M**
usherin		
2299delG	0.995937	N/N

Figure 9